



THE CENTRAL AFRICAN JOURNAL OF MEDICINE

Vol. 40, No.12

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Details of instructions to authors are published in the January and July issues of the Journal.

Manuscripts submitted for publication are accepted on the understanding that they are contributed exclusively to the *The Central African Journal of Medicine*. A statement to that effect should be included in the letter accompanying the manuscript.

Communications concerning editorial matter, advertising, subscription, change of address, etc., should be addressed to the Administrative Manager, P.O. Box A 195, Avondale, Harare, Zimbabwe.

The subscription rate (including surface postage) for 1995 is Z\$140.00 per annum locally; Europe US\$110.00, Africa US\$100.00 and US\$110.00 elsewhere. The subscription rate (including airmail postage) for 1995 in Africa is US\$175.00; Europe US\$195.00 and US\$210.00 elsewhere.

Owned and published by The Central African Journal of Medicine Company in conjunction with the University of Zimbabwe Faculty of Medicine.

Delayed diagnosis of retinoblastoma

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SUMMARY

The initial clinical presentation of retinoblastoma can mimic other non-malignant conditions. This may cause a delay in accurate diagnosis with fatal consequences.

Two cases are presented in which the initial impression of panophthalmitis and congenital cataract, respectively, led to delay in diagnosis. The need for early clinical diagnosis bearing in mind the masquerading tendency of retinoblastoma is emphasized. Treatment modalities readily available in our environment are highlighted.

INTRODUCTION

Retinoblastoma is the commonest childhood intra-ocular malignancy.¹ Kodilinye² first showed that the disease is not uncommon in Nigeria, when he reported an occurrence rate of one in 192 new patients in Ibadan. His series recorded a high mortality rate which was attributed mainly to late presentation. Abiose³ in an analysis of 500 consecutive children seen over a six month period at Kaduna, Northern Nigeria, found that 1.2 pc had retinoblastoma. A study in Zaire,⁴ noted that delayed diagnosis was responsible for higher mortality in retinoblastoma. Reasons adduced for delayed diagnosis include lack of knowledge, lack of financial means and the long distance most families are required to travel for eye care.

Data from the United States of America indicate that retinoblastoma occurs in one in 14 000 to 20 000 live births and that there are 250 to 500 new cases each year.¹

Although its commonest symptoms and signs are leukokoria (white pupil), strabismus and visual loss,¹

the disease can also present in other forms which clinically may be confused with such ocular disorders as panophthalmitis, endophthalmitis, congenital cataract or phthisis bulbi.⁵ These masquerading symptoms and signs of retinoblastoma may cause a delay in accurate diagnosis with fatal consequences.

This communication reports two cases of retinoblastoma which were initially diagnosed and managed for sometime as panophthalmitis and congenital cataract, respectively.

CASE REPORTS

Case 1: SO was a one year 10 months old boy who was brought to the children's outpatient clinic of our teaching hospital on 11 September 1991, with a history of recurrent swelling and redness of the right eye. The latest episode started three days prior to presentation.

When he was four months old, the mother had noticed a white patch in the patient's right eye. About a month later the eye became red and swollen. She took the child to an eye hospital where he was treated until the swelling subsided. A few months later, the swelling recurred and this time the patient was referred to a teaching hospital in the area. The patient was once more treated conservatively and when the symptoms subsided, he was discharged and given a six month follow up appointment. Symptoms recurred again before the appointment date.

At this stage, the patient was brought to the children's outpatient clinic of our teaching hospital where a clinical impression of ophthalmitis was made. A ocular malignancy was also considered. The patient was admitted and ophthalmic opinion sought.

The ophthalmologist made a diagnosis of retinoblastoma of the right eye. On 17 September 1991 the patient had enucleation of the right eye and examination under anaesthesia (UEA) of the left eye was normal while histopathology of the enucleated right eye confirmed the diagnosis of retinoblastoma. The tumour was necrotic in some areas. The patient was referred for radiotherapy.

Case 2: PT was a three year old boy who presented at the Guinness Eye Centre, Onitsha, on 26 March 1992 with a swollen right eye.

When he was three weeks old, the mother had noticed a white patch in the black part of the right eye. She took the child to a private eye hospital where she was told the child had cataract and would be operated upon when he was six months old. At the age of six

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months, the surgery was not performed although the patient's parents were ready for it.

The black part of the eye remained white until a few weeks prior to presentation at our hospital, when the right eye became swollen.

Ocular examination of the right eye showed swollen eye lids, proptosis, marked conjunctival injection, hazy cornea and hypopyon. The left eye apparently was normal. An impression of retinoblastoma was made.

On 3 April 1992, the patient had enucleation of the right eye and examination under anaesthesia (EUA) of the left. The left eye was normal. Histopathology report on the enucleated eye confirmed a diagnosis of retinoblastoma with involvement of the optic nerve (Figure 1). The patient was referred for radiotherapy.

Figure 1: Child (Case 2) with advanced retinoblastoma in the right eye (which had been managed as panophthalmitis).



DISCUSSION

Early diagnosis of retinoblastoma is important not only because it is a life threatening disease, but also because a delay in diagnosis affects treatment options and prognosis.⁶ Treatment for patients presenting with the advanced disease is essentially palliative.⁷ But a rational application of radiotherapy and/or other treatment modalities, for cases diagnosed early, ensured a better prognosis for vision and longevity for the patients.

About a century ago, retinoblastoma was uniformly fatal even in developed countries. But early diagnosis and application of modern treatment methods have improved the survival rate which now ranges between 80 and 90 pc in developed countries.⁸⁻¹⁰

This profound success can only be achieved where patients present early and accurate diagnosis is made.

Late presentation to a hospital is usually advanced as part of the reason for the dismal outcome of retinoblastoma and other eye diseases in developing countries.^{2,4} However, with increasing education of the populace and the availability of modern health care facilities within the reach of many people, the trend is changing.

In the two cases herein reported, the parents sought medical help for the children early at the appropriate health institutions. That accurate diagnosis in both cases were delayed for so long, underscores the need to once more highlight some of the masquerading symptoms and signs of retinoblastoma.

In the majority of cases, the diagnosis of retinoblastoma can be made on clinical grounds alone.^{1,6} The key to diagnosis is a thorough ocular examination which in a child may require examination under anaesthesia (EUA). Detailed and careful assessment of each eye, which includes indirect ophthalmoscopy will in most cases reveal evidence of a tumour. Such examination will also show that leukokoria is actually due to a retrolental mass, and quite different from lens opacity encountered in congenital cataract.

The physician should be suspicious of any case of recurrent ocular and periocular inflammation especially in children. Up to two pc of retinoblastoma patients can present with a painful red eye; a similar proportion also presented with orbital inflammation and proptosis.

The intraocular tumour in retinoblastoma can undergo necrosis as the histopathology report in one of our cases showed. The dead tissue excites inflamma-

tory reactions that can masquerade as endophthalmitis or panophthalmitis. In cases of doubt, cytology of an aqueous humour tap can aid diagnosis.⁶

Enucleation, chemotherapy, irradiation, cryotherapy and photocoagulation are treatment options for retinoblastoma.⁶ Enucleation and chemotherapy can be readily practised by the average ophthalmologist or in conjunction with a paediatrician interested in oncology. However, chemotherapy is expensive and this coupled with possible scarcity of drugs in developing countries, may lead to poor compliance. Cultural taboos could also cause a rejection of enucleation. But contrary to the experience of others,⁵ the parents of the children herein reported never hesitating when enucleation was offered.

Kodilinye² in 1987 lamented the lack in Nigeria of such treatment as radiotherapy. Some developing countries, including Nigeria, now have functioning radiotherapy units. The opportunity offered by this facility should be maximised through early diagnosis and quick referral of retinoblastoma patients.

Finally, the awareness of masquerading symptoms and signs; earlier diagnosis and the availability of modern modes of therapy, expectedly should improve the rate of survival of retinoblastoma patients even in the developing countries.

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